

Organisms (Continuity and change) revision notes

How organisms reproduce, pass on their genes and keep their internal conditions stable. D3.1 covers sexual and asexual reproduction in humans and flowering plants, D3.2 covers the patterns of inheritance, and D3.3 covers homeostasis, from blood glucose to body temperature.

For IB Diploma Biology SL undefined. **HL only** is additional higher level content, for HL students only.

1 Reproduction Spec D3.1

Sexual and asexual reproduction Spec D3.1.1, D3.1.2

In **asexual reproduction** one parent produces offspring by mitosis (or a similar division). The offspring are **genetically identical** to the parent and to each other, apart from rare mutations.

In **sexual reproduction** two parents each produce haploid gametes. The gametes fuse, so each offspring has a **new combination of genes** that is different from both parents and from its siblings.

Relative advantages of the two methods		
Feature	Asexual reproduction	Sexual reproduction
Number of parents	One	Usually two (or one hermaphrodite producing both gametes)
Genetic outcome	Offspring genetically identical to the parent	Offspring with new gene combinations, so genetic variation
When it is an advantage	Parent is already adapted to an existing, stable environment, so its successful combination of alleles is kept	Environment changes: variation gives some offspring traits suited to the new conditions, so the population can adapt
Other points	Fast; no need to find a mate; every individual can produce offspring	Slower; needs gametes from two parents to meet

Meiosis and fusion of gametes in the sexual life cycle

A sexual life cycle alternates between a diploid and a haploid stage. **Meiosis** halves the chromosome number to produce haploid gametes, and **fusion of gametes** (also called fertilization) restores the diploid number in the zygote.

- **Meiosis breaks up parental combinations of alleles:** crossing over and random orientation of bivalents mean each gamete carries a unique mix of the parent's maternal and paternal alleles.
- **Fusion of gametes produces new combinations:** a gamete from one parent joins a gamete from another, so the zygote combines two different sets of alleles.
- Without meiosis the chromosome number would double every generation.

Exam tip: When asked for an advantage, tie it to the environment: asexual reproduction for an existing environment the parent is adapted to, sexual reproduction for a changed environment.

Male and female sexes and human reproductive systems Spec D3.1.3, D3.1.4

The **prime difference** between the sexes is that the **male gamete travels to the female gamete**. A moving cell must be small, so sperm have very little cytoplasm and almost no food reserves. The egg

does not move, so it can be large and store food reserves for the early embryo.

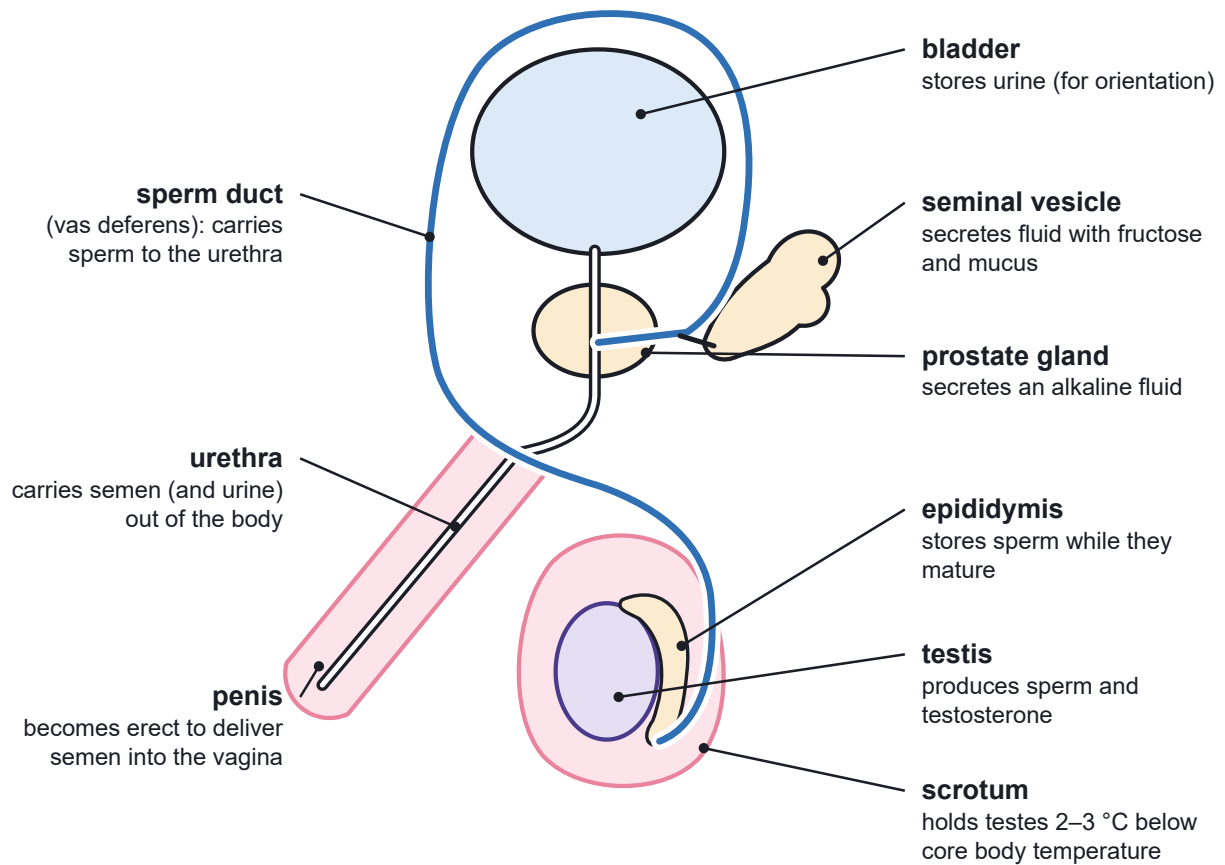
- **Numbers of gametes:** small gametes are cheap to make, so males produce huge numbers (millions of sperm per ejaculation); females produce few, large eggs (usually one per menstrual cycle in humans).
- **Reproductive strategies:** because eggs are scarce and costly, females usually invest more in each offspring (here, pregnancy and lactation); males can potentially father many offspring and often compete for mates.

Male reproductive system

Structures and functions: male-typical system	
Structure	Function
Testis	Produces sperm in seminiferous tubules and secretes testosterone
Scrotum	Holds the testes outside the body, about 2-3 °C below core temperature, which sperm production needs
Epididymis	Stores sperm while they mature and gain the ability to swim
Sperm duct (vas deferens)	Carries sperm from the epididymis towards the urethra during ejaculation
Seminal vesicle	Secretes fluid containing fructose (energy for sperm) and mucus into semen
Prostate gland	Secretes an alkaline fluid that helps neutralise acidity in the vagina
Urethra	Carries semen (and urine) out through the penis
Penis	Becomes erect to deliver semen into the vagina

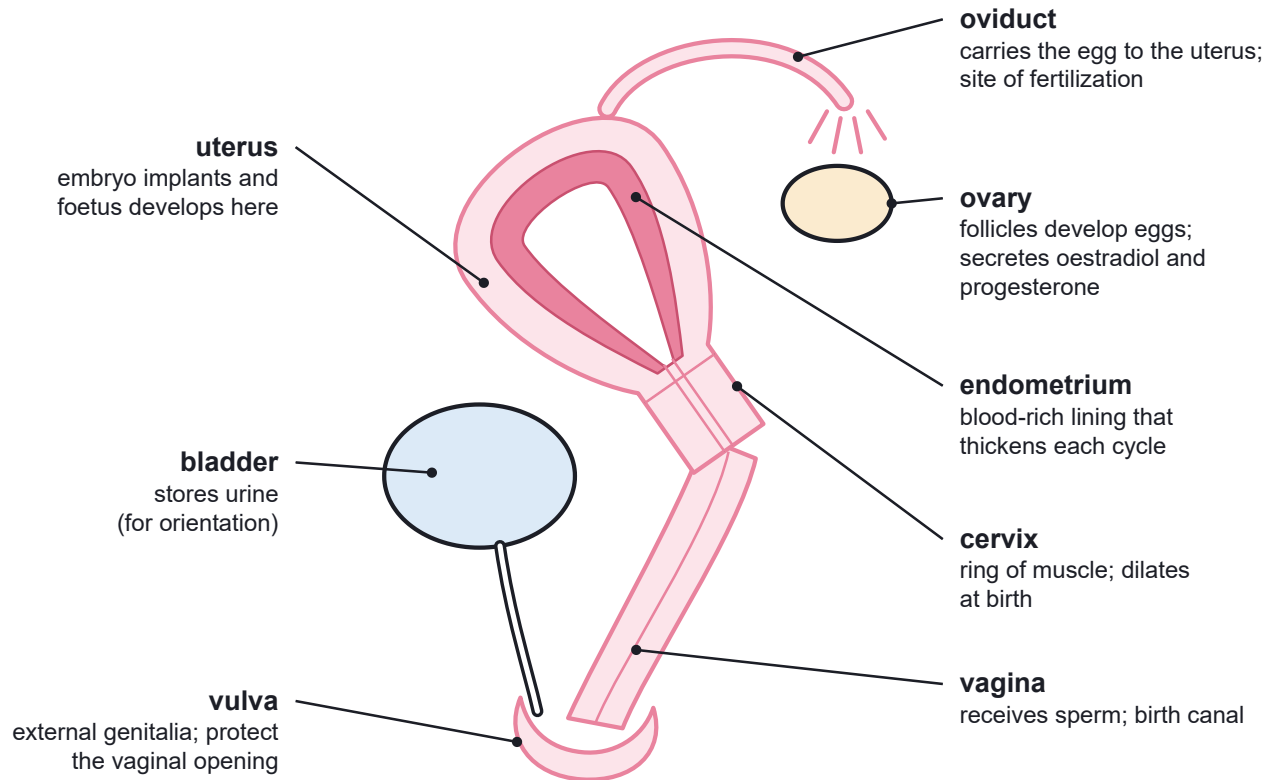
Female reproductive system

Structures and functions: female-typical system	
Structure	Function
Ovary	Contains follicles that develop eggs; releases an egg at ovulation; secretes oestradiol and progesterone
Oviduct (fallopian tube)	Carries the egg towards the uterus; site of fertilization
Uterus	Muscular organ in which the embryo implants and the foetus develops
Endometrium	Blood-rich lining of the uterus that thickens each cycle, ready for implantation
Cervix	Ring of muscle at the base of the uterus; keeps the foetus in place and dilates at birth
Vagina	Receives the penis and sperm; the birth canal
Vulva	External genitalia that protect the vaginal opening



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The male-typical reproductive system (side view, front of the body on the left). Sperm made in the **testis** mature in the epididymis and leave through the sperm duct and urethra.



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The female-typical reproductive system (side view, front of the body on the left). The egg is fertilized in the **oviduct** and the embryo implants in the endometrium.

Exam tip: Drawing questions want labels with functions. Draw clear single lines, use a ruler for label lines and write the function next to each structure.

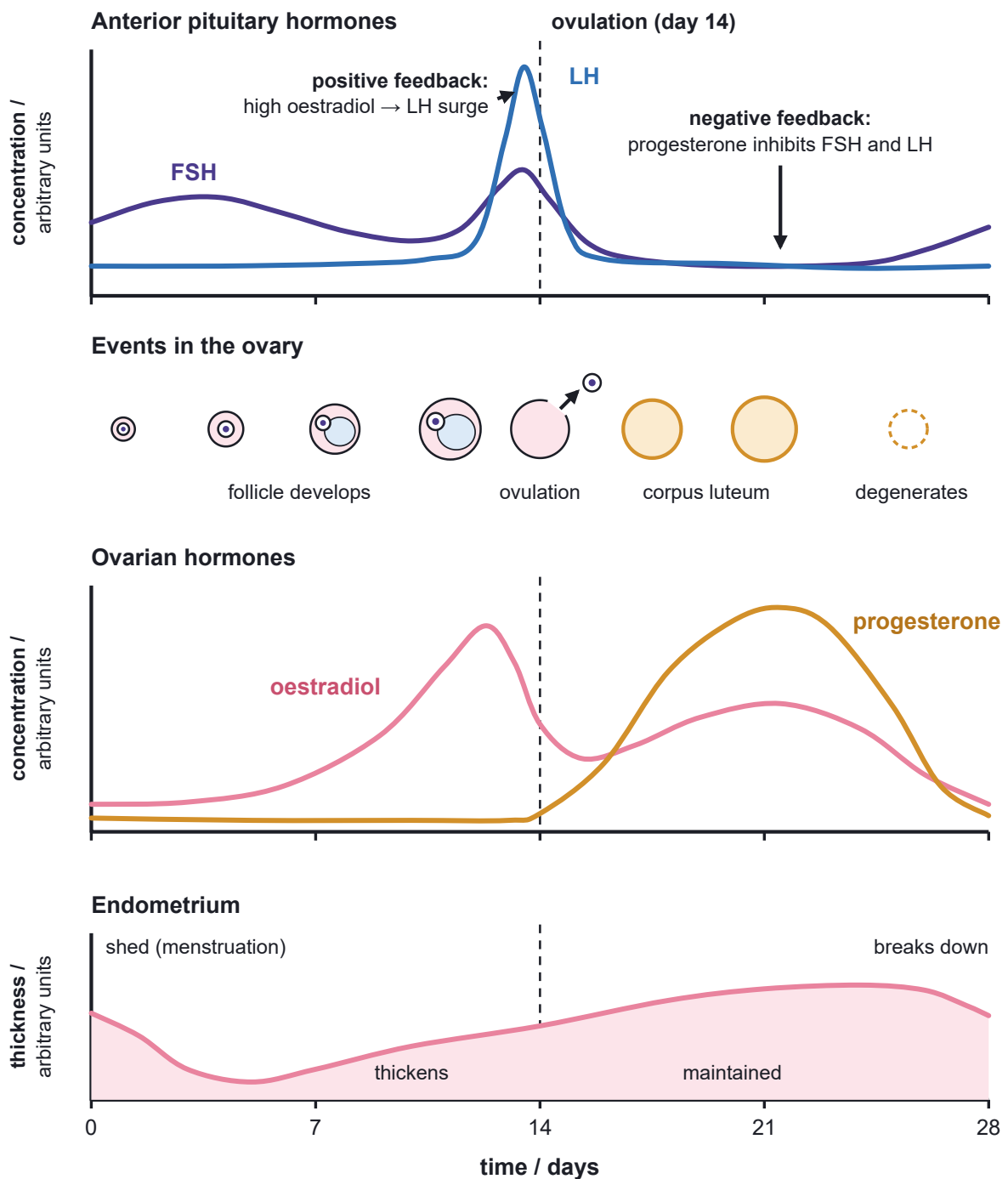
Ovarian and uterine cycles and their hormones Spec D3.1.5

The **ovarian cycle** (follicle growth, ovulation, corpus luteum) and the **uterine cycle** (endometrium builds up, is maintained, then breaks down) together make the **menstrual cycle**, roughly 28 days long.

The four hormones		
Hormone	Secreted by	Main effects
FSH (follicle-stimulating hormone)	Anterior pituitary	Stimulates development of follicles; stimulates follicle cells to secrete oestradiol
LH (luteinizing hormone)	Anterior pituitary	Surge triggers ovulation; stimulates the empty follicle to become the corpus luteum
Oestradiol	Follicle cells in the ovary	Thickens the endometrium; low levels inhibit FSH and LH; high levels stimulate the LH and FSH surge
Progesterone	Corpus luteum	Maintains and further thickens the endometrium; inhibits FSH and LH secretion

1. Days 1–5: the endometrium is shed (menstruation). Progesterone and oestradiol are low, so FSH secretion rises.

- 2 . FSH stimulates several follicles to develop; usually one matures. Its cells secrete increasing amounts of oestradiol.
- 3 . Oestradiol stimulates the endometrium to thicken again. At first it inhibits FSH (negative feedback).
- 4 . By about day 12-13 oestradiol is high. High oestradiol stimulates the pituitary to release much more LH and FSH: **positive feedback** causes the LH surge.
- 5 . The LH surge triggers ovulation at about day 14: the mature follicle releases a secondary oocyte into the oviduct.
- 6 . LH stimulates the empty follicle to develop into the **corpus luteum**, which secretes progesterone (and some oestradiol).
- 7 . Progesterone maintains the thick endometrium and, with oestradiol, inhibits FSH and LH (**negative feedback**), so no new follicles develop.
- 8 . If there is no pregnancy, the corpus luteum degenerates after about 10-12 days. Progesterone falls, the endometrium breaks down (menstruation) and FSH rises again: the cycle repeats.



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The menstrual cycle. High oestradiol causes the LH surge by **positive feedback**; progesterone from the corpus luteum inhibits FSH and LH by **negative feedback**.

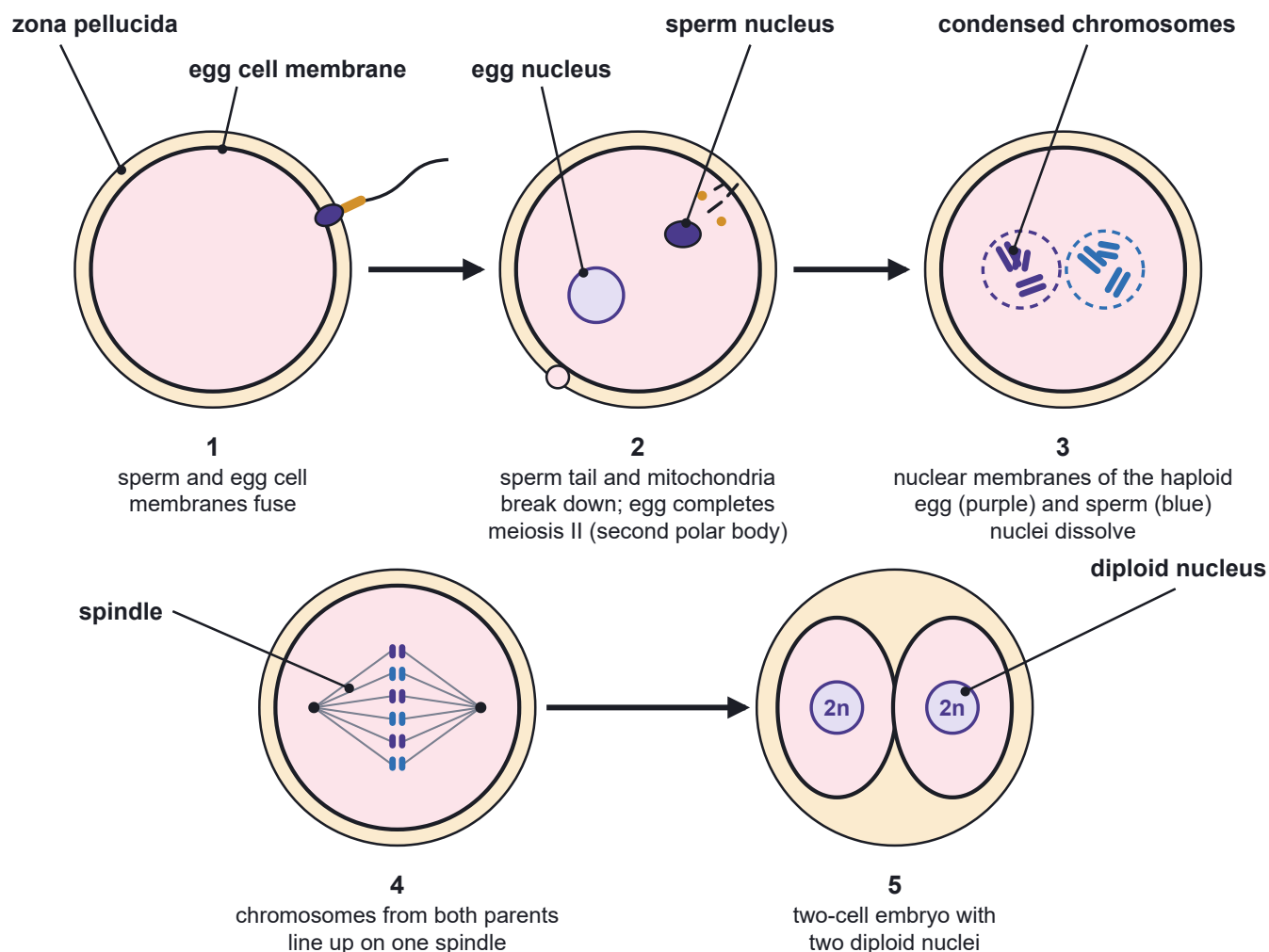
Common mistake: Oestradiol has both effects: low to moderate levels inhibit FSH and LH, but sustained high levels before ovulation stimulate the LH surge. Say which level you mean.

Fertilization in humans and hormones in IVF Spec D3.1.6, D3.1.7

Fertilization

1. Fertilization usually happens in the oviduct. A sperm reaches the egg and its **cell membrane fuses with the egg cell membrane**.
2. The sperm nucleus enters the egg cytoplasm. The sperm's **tail and mitochondria are destroyed**, so mitochondria (and mitochondrial DNA) are inherited from the mother only.

3. The egg, a secondary oocyte, completes its second meiotic division, so a haploid egg nucleus and a haploid sperm nucleus are both present in the cytoplasm.
4. The **nuclear membranes of the sperm and egg nuclei dissolve**; the two nuclei do not fuse into one nucleus first.
5. All the condensed chromosomes, from both parents, take part in a **joint mitosis** on one spindle. This produces **two diploid nuclei**, so the zygote divides into a two-cell embryo.



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Fertilization in humans. The two haploid nuclei do not fuse first: their chromosomes join in a **joint mitosis** on one spindle, giving two diploid nuclei.

Hormones in IVF

In **in vitro fertilization (IVF)** eggs are fertilized outside the body. Hormones are used so that many eggs can be collected at once.

1. **Down-regulation:** a drug is given (usually daily for a few weeks) that stops the pituitary releasing FSH and LH, so the **normal secretion of hormones is suspended** and the woman's own cycle cannot interfere with the timing.
2. **Superovulation:** injections of FSH (with LH activity) are given at a much higher dose than normal, so many follicles develop at once.
3. When follicles are mature, an injection of hCG (which acts like LH) makes the eggs complete their maturation; they are collected from the follicles about 36 hours later.

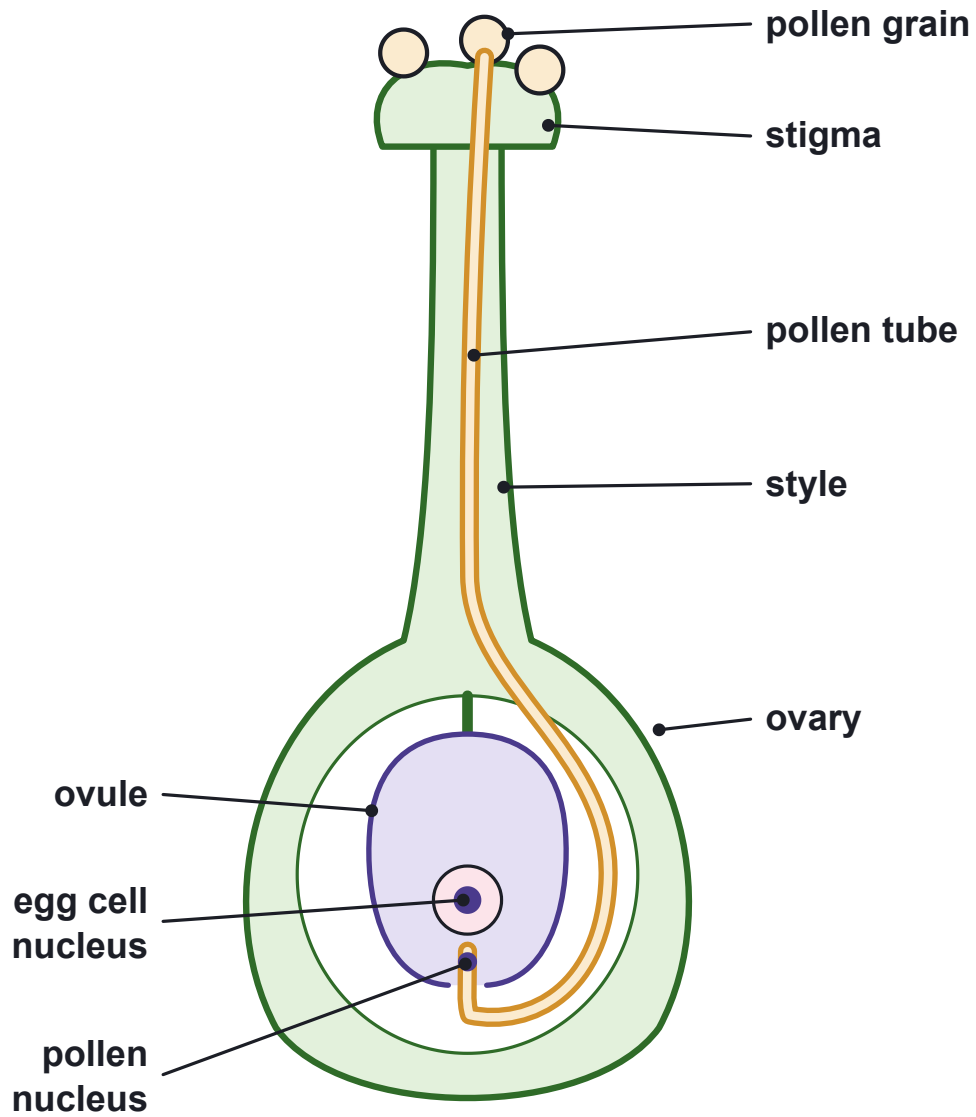
4. Eggs are mixed with sperm (or a sperm is injected into each egg) and the embryos are cultured for a few days.
5. One or two embryos are transferred to the uterus. Progesterone is given to help keep the endometrium ready for implantation.

Exam tip: For IVF, use the two key phrases: normal secretion of hormones is suspended, then artificial doses of hormones induce superovulation.

Sexual reproduction in flowering plants and insect-pollinated flowers Spec D3.1.8, D3.1.9

Flowering plants reproduce **sexually**, even when one plant (a hermaphrodite) has both male and female parts, because haploid gametes still fuse at fertilization.

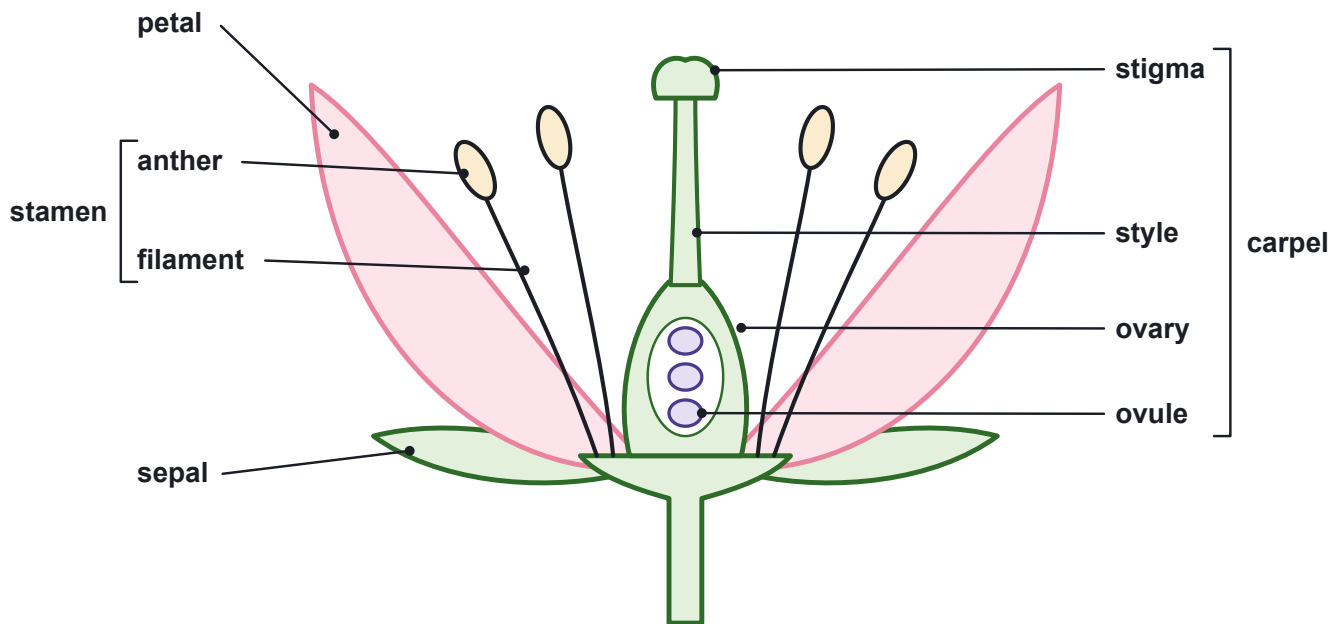
1. **Gamete production:** male gametes form inside **pollen grains** made in the anthers; the female gamete (egg cell) forms inside each **ovule** in the ovary.
2. **Pollination:** pollen is transferred from an anther to a stigma, by animals or wind.
3. **Pollen development:** the pollen grain absorbs water and sugar on the stigma and germinates. A pollen tube grows down through the style to the ovary, carrying the male gametes.
4. **Fertilization:** the tube enters an ovule through the micropyle and a male gamete nucleus fuses with the egg cell nucleus, giving a diploid zygote.
5. The zygote divides by mitosis to form an **embryo**. The ovule becomes a seed and the ovary develops into a fruit.



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After pollination the **pollen tube** grows down the style and delivers the male gametes to the ovule. The 'pollen nucleus' in the figure is the male gamete nucleus that fuses with the egg cell nucleus.

Features of an insect-pollinated flower



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An insect-pollinated flower. The **stamen** (anther and filament) is male; the **carpel** (stigma, style, ovary, ovule) is female.

Structures and functions	
Structure	Function
Petal	Large, brightly coloured and often scented to attract insects
Nectary	Secretes nectar, a sugary reward for insect visitors
Anther	Produces pollen; placed where a visiting insect brushes against it
Filament	Stalk holding the anther in position
Stigma	Sticky surface that receives pollen from an insect
Style	Holds the stigma up; the pollen tube grows through it
Ovary	Contains the ovules; becomes the fruit
Ovule	Contains the female gamete; becomes the seed
Sepal	Protects the flower while it is a bud

Exam tip: Annotated diagrams score for function, not just names: write what each structure does next to its label, as in the table.

Cross-pollination and self-incompatibility Spec D3.1.10, D3.1.11

Self-pollination (pollen reaching a stigma of the same plant) leads to **inbreeding**. Offspring become more homozygous, so genetic diversity falls and harmful recessive alleles are expressed more often, reducing vigour. **Cross-pollination** transfers pollen to a different plant of the same species.

Methods of promoting cross-pollination

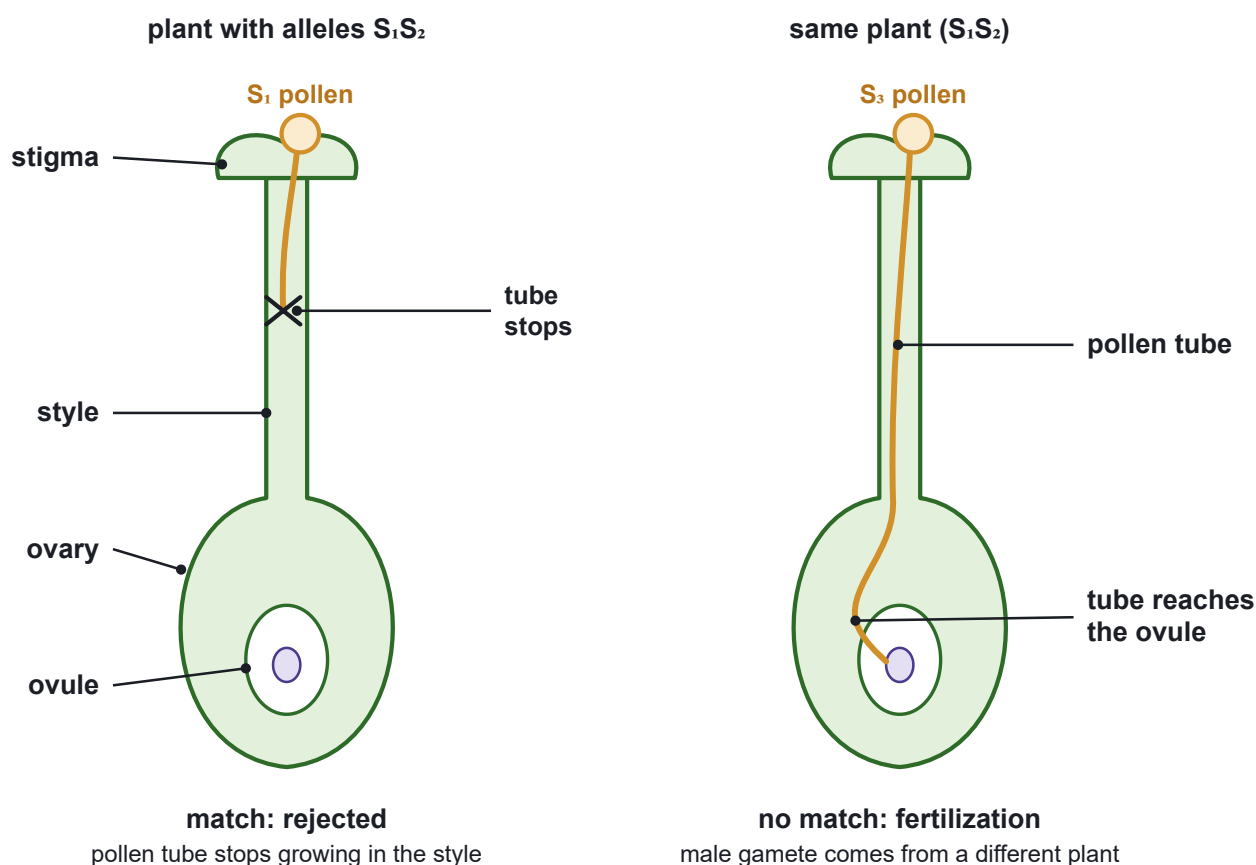
- **Different maturation times:** the anthers release pollen before the stigma of the same flower is receptive (or the other way round), so pollen has to come from another flower.
- **Separate male and female flowers** on the same plant, for example maize, with male flowers at the top and female flowers lower down.

- **Separate male and female plants**, for example holly: a female plant can only be pollinated by pollen from a male plant.
- **Animals or wind** carry pollen between plants: insects, birds and bats travel from flower to flower, and wind carries light pollen long distances.

Self-incompatibility

In many species a **self-incompatibility** gene (often called the S gene) has many alleles. In the commonest type, if the S allele carried by a pollen grain matches either S allele of the plant it lands on, the pollen is recognised as 'self' and its pollen tube stops growing in the style. In some other species, such as the cabbage family, the pollen's S type is set by the plant that made it, and matching pollen fails to germinate on the stigma.

- Pollen from the same plant always matches, so self-fertilization is blocked.
- Pollen from a plant with different S alleles grows normally, so male and female gametes that fuse come from different plants.
- This keeps genetic variation within the species high.



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Self-incompatibility. Pollen whose S allele matches either allele of the plant is rejected, so the male gamete must come from a different plant.

Exam tip: Linking question: how can interspecific relationships assist reproductive strategies? Animal pollinators gain nectar or pollen as food while carrying pollen between plants.

Seed dispersal and germination Spec D3.1.12

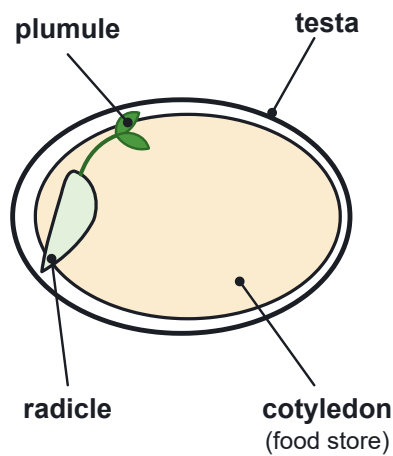
Distinguishing pollination and seed dispersal		
	Pollination	Seed dispersal
What moves	Pollen (containing male gametes)	Seeds (containing embryos), often inside fruits
From and to	Anther to stigma	Parent plant to a new site away from it
When	Before fertilization	After fertilization, once seeds are mature
Purpose	Allows fertilization	Reduces competition with the parent and siblings; spreads the species to new areas

- **Wind:** light seeds or fruits with wings or parachutes (sycamore, dandelion).
- **Animals:** fleshy fruits eaten and seeds passed out in faeces; hooked fruits that catch on fur.
- **Water:** buoyant fruits such as coconuts.
- **Explosive:** pods that dry and burst, flinging seeds (peas and other legumes).

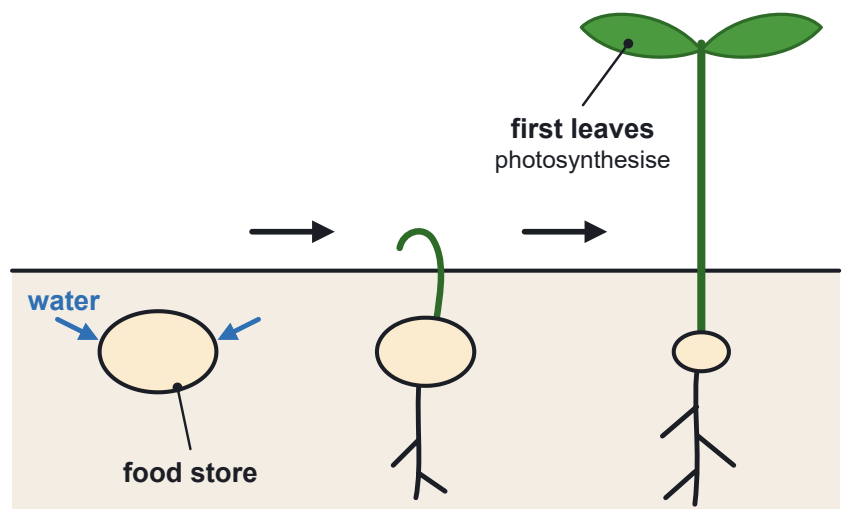
Germination

A seed contains an embryo (radicle, plumule and cotyledons) and food reserves, inside a protective testa. Germination needs **water, oxygen and a suitable temperature**.

1. The seed absorbs water, so it swells, the testa splits and enzymes are activated.
2. The embryo produces the plant growth regulator gibberellin (gibberellic acid), which stimulates production of hydrolytic enzymes such as amylase. This is best understood in cereal grains, where the enzymes are made in the aleurone layer around the food store.
3. Food reserves are **mobilized**: amylase digests starch into maltose and sugars, proteases digest proteins into amino acids, and lipases digest lipids.
4. These soluble products are transported to the growing regions of the embryo for aerobic respiration and growth.
5. The radicle grows out first and becomes the root; the plumule grows up into the shoot. Once leaves open and photosynthesise, the seedling no longer depends on its reserves.



Inside a bean seed



1 Seed takes in water and swells

2 Root grows down, shoot grows up

3 Seedling with green leaves

Food reserves are used until photosynthesis begins

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A bean seed and its germination. Food reserves in the cotyledons are used until **photosynthesis** begins.

Common mistake: Do not mix up dispersal and pollination: pollination moves pollen before fertilization; dispersal moves seeds after it.

Quick check

1. State one advantage of asexual reproduction.
2. Which hormone surge triggers ovulation, and what causes it?
3. What happens to the sperm's tail and mitochondria at fertilization?
4. What is the function of the stigma?

2 Inheritance Spec D3.2

Haploid gametes, diploid zygotes and genetic crosses in plants Spec D3.2.1, D3.2.2

Each parent produces **haploid** gametes by meiosis, carrying **one copy of each gene**. At fertilization two gametes fuse to give a **diploid** zygote with **two copies of each autosomal gene**, one from each parent. This pattern of inheritance is common to all eukaryotes with a sexual life cycle.

Carrying out a cross in flowering plants

Pollen contains the male gametes and the female gametes are in the ovules inside the ovary, so **pollination is needed to carry out a cross**. Plants such as peas produce male and female gametes in the same flower, so they normally **self-pollinate** and self-fertilize.

1. Choose two pure-breeding (homozygous) parents with contrasting traits: the **P generation**.
2. Remove the immature anthers from flowers of one parent so it cannot self-pollinate.
3. Transfer pollen from the other parent to its stigmas with a small brush, then cover the flowers with a bag to stop other pollen arriving.
4. Collect the seeds and grow them: these plants are the **F1 generation**. Record their phenotypes.
5. Allow the F1 plants to self-pollinate (or cross them with each other). The offspring are the F2 generation. Count each phenotype.
6. Use a **Punnett grid** to predict the expected ratios and compare them with the counts.

Genetic crosses like these are widely used to breed new varieties of crop and ornamental plants, combining useful traits such as yield, disease resistance or flower colour.

Exam tip: Always show a full genetic diagram: parental phenotypes and genotypes, gametes, a Punnett grid, then offspring genotypes, phenotypes and ratio.

Genotype, phenotype, dominance and phenotypic plasticity Spec D3.2.3, D3.2.4, D3.2.5, D3.2.6

Gene	A heritable factor that occupies a specific position (locus) on a chromosome and influences a characteristic.
Allele	One of the alternative forms of a gene, differing in base sequence by one or a few bases.
Genotype	The combination of alleles of a gene (or genes) inherited by an organism.
Homozygous	Having two identical alleles of a gene, for example AA or aa.
Heterozygous	Having two different alleles of a gene, for example Aa.
Phenotype	The observable traits of an organism, resulting from its genotype and environmental factors.

Causes of phenotype in humans	
Cause	Examples
Genotype only	ABO blood group; inherited conditions such as haemophilia
Environment only	Scars, tattoos, the language a person speaks
Interaction of genotype and environment	Height and body mass (genes plus nutrition); skin colour (genes plus exposure to sunlight)

Dominant and recessive alleles

A **dominant allele** usually codes for a functional protein, often an enzyme. One copy produces enough of it for the trait to appear, so **homozygous dominant (AA) and heterozygous (Aa) individuals have the same phenotype**. A **recessive allele** often codes for a non-functional protein, or none, so its effect only shows when there is no dominant allele (aa).

Phenotypic plasticity

Phenotypic plasticity is the capacity to develop traits suited to the environment an organism experiences, by **varying patterns of gene expression**. The genotype does not change, and the changes may be **reversible** during the individual's lifetime.

- Human skin tans in strong sunlight as melanin production increases, then fades in winter.
- Skeletal muscles grow larger with regular training and shrink when training stops.
- Water crowfoot (*Ranunculus aquatilis*) grows finely divided leaves under water and broad leaves above it, from the same genotype.

Common mistake: Phenotypic plasticity is not mutation or evolution. The alleles stay the same; only which genes are expressed, and how much, changes.

Phenylketonuria, SNPs, multiple alleles and ABO blood groups Spec D3.2.7, D3.2.8, D3.2.9

Phenylketonuria

Phenylketonuria (PKU) is a recessive genetic condition caused by a mutation in an **autosomal gene** that codes for the enzyme phenylalanine hydroxylase, needed to **convert phenylalanine to tyrosine**.

- Only people with two recessive alleles (pp) have PKU. Heterozygotes (Pp) make enough enzyme from their one functional allele, so they are unaffected carriers.
- Without the enzyme, phenylalanine from dietary protein builds up in the blood and damages the developing brain, causing intellectual disability. Less tyrosine is made, so less melanin.
- Babies are screened soon after birth with a blood test. Affected children follow a diet low in phenylalanine, which prevents most harm: an example of environment affecting phenotype.

Single-nucleotide polymorphisms and multiple alleles

A **single-nucleotide polymorphism (SNP)** is a position in a gene where the base differs between individuals. Each new SNP arising by mutation can create a new allele, so **any number of alleles** of a gene can exist in the **gene pool** (all the alleles in an interbreeding population). A diploid individual, though, only inherits **two** alleles of each autosomal gene.

ABO blood groups

The ABO gene has three common alleles. **I^A** and **I^B** code for enzymes that add different sugars to a glycoprotein antigen on red blood cells; **i** codes for a non-functional enzyme. I^A and I^B are both dominant to i, and codominant with each other.

ABO genotypes and phenotypes		
Blood group	Genotypes	Antigen on red blood cells
A	I ^A I ^A or I ^A i	A
B	I ^B I ^B or I ^B i	B
AB	I ^A I ^B	A and B
O	ii	Neither

An ABO cross

A father of blood group A ($I^A i$) and a mother of blood group B ($I^B i$) have a child. Determine the possible blood groups of their children and the probability of each.

1. Father's gametes: I^A or i . Mother's gametes: I^B or i .
2. Punnett grid: $I^A I^B$, $I^A i$, $I^B i$, ii .
3. Phenotypes: AB, A, B, O, one of each.

Answer: Each child has a 0.25 (25%) chance of each blood group: A, B, AB or O.

Exam tip: Use exactly the notation I^A , I^B and i for ABO alleles. Writing A, B and O as alleles usually loses the mark.

Incomplete dominance and codominance Spec D3.2.10

Patterns of inheritance at the phenotypic level

Pattern	Heterozygote phenotype	Example
Complete dominance	Same as homozygous dominant	Pea plants: Tt is tall, like TT
Incomplete dominance	Intermediate between the two homozygotes	Four o'clock flower (<i>Mirabilis jalapa</i>): red × white gives pink
Codominance	Dual phenotype: both alleles expressed	Blood group AB ($I^A I^B$): both A and B antigens present

Incomplete dominance in the four o'clock flower

A pure-breeding red *Mirabilis jalapa* ($C^R C^R$) is crossed with a pure-breeding white plant ($C^W C^W$). The F1 are self-pollinated. Deduce the F1 and F2 phenotypes and ratio.

1. P gametes: C^R and C^W , so all F1 are $C^R C^W$: pink.
2. F1 × F1: each produces C^R and C^W gametes in equal numbers.
3. Punnett grid: $C^R C^R$, $C^R C^W$, $C^W C^R$, $C^W C^W$.

Answer: F1 all pink. F2 1 red : 2 pink : 1 white; the genotype ratio and phenotype ratio are the same.

In the pink heterozygote, the single C^R allele produces only enough red pigment to make the petals pink. In blood group AB, each allele makes its own enzyme, so both antigens appear side by side.

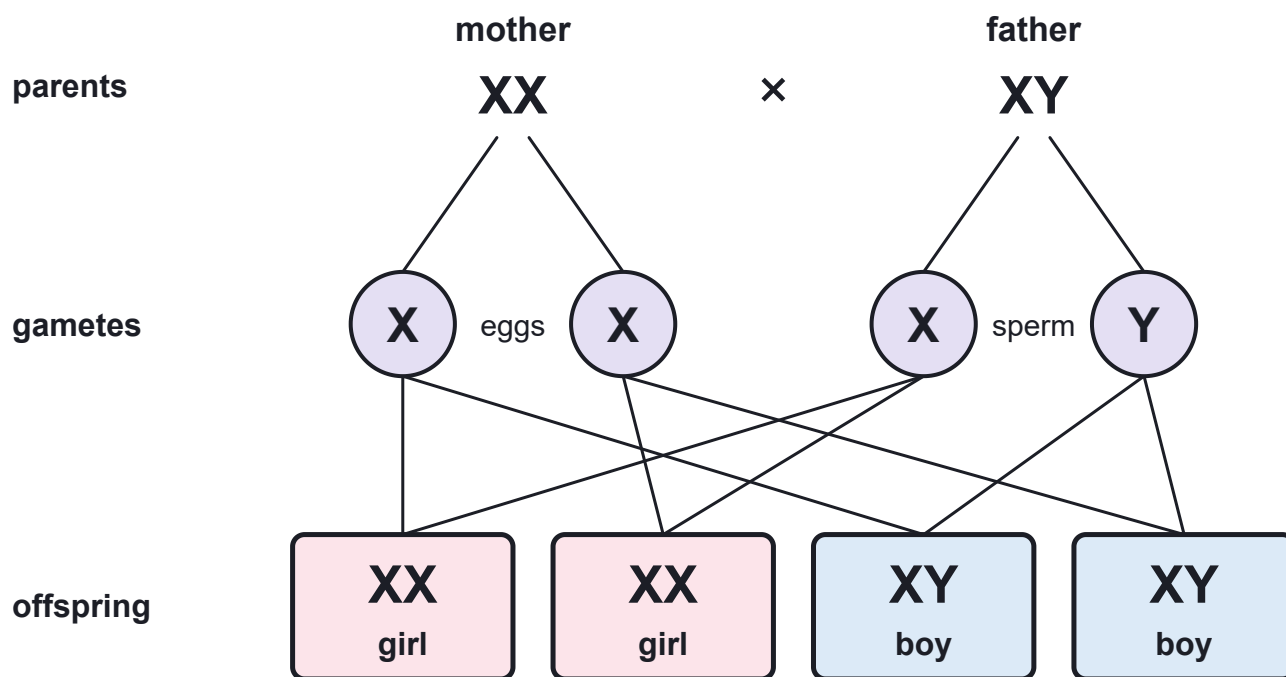
Exam tip: A 1:2:1 phenotype ratio in an F2 is a sign of incomplete dominance or codominance, because the heterozygotes can be told apart. Either the common or the scientific name of the organism is acceptable.

Sex determination, sex linkage and haemophilia Spec D3.2.11, D3.2.12

Human body cells have 22 pairs of autosomes and one pair of **sex chromosomes**: **XX** in female-typical and **XY** in male-typical individuals. Every egg carries an X. Half the sperm carry an X and half a Y, so **the sex chromosome in the sperm determines** the development of the zygote.

- A gene on the Y chromosome (SRY) causes the embryonic gonads to develop into testes. Testosterone from the testes then leads to male-typical characteristics. Without it, ovaries and female-typical characteristics develop.
- The X chromosome is much larger and carries **far more genes** than the Y chromosome, most of them nothing to do with sex.

- Genes carried on the X chromosome are **sex-linked**. A male has only one X, so whichever allele he has on it is expressed, even if recessive.



2 girls : 2 boys, a ratio of 1 : 1

50% chance of a girl and 50% chance of a boy

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Inheritance of sex: the sperm carries X or Y, so each child has a **50% chance** of XX (female-typical) or XY (male-typical).

Haemophilia

In **haemophilia** the blood clots very slowly because a clotting protein (factor VIII in the commonest type) is missing. The gene is on the X chromosome. The normal allele is **X^H** and the recessive haemophilia allele **X^h**.

Haemophilia genotypes	
Genotype	Phenotype
X^HX^H	Unaffected female
X^HX^h	Unaffected carrier female
X^hX^h	Affected female (rare)
X^HY	Unaffected male
X^hY	Affected male

Carrier mother and unaffected father

A carrier woman ($X^H X^h$) and an unaffected man ($X^H Y$) have children. Deduce the probabilities of the possible outcomes.

1. Mother's gametes: X^H or X^h . Father's gametes: X^H or Y .
2. Punnett grid: $X^H X^H$, $X^h X^H$, $X^H Y$, $X^h Y$.
3. Daughters: half unaffected, half carriers. Sons: half unaffected, half affected.

Answer: Probability a child is an affected son = 0.25; probability that a son is affected = 0.5; no daughters affected.

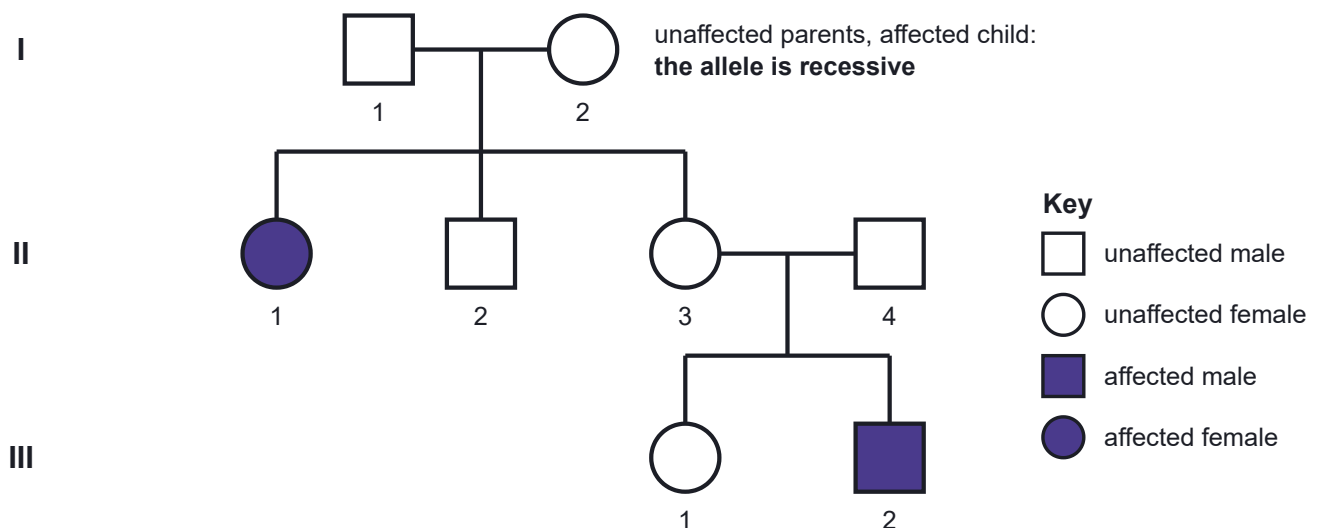
Common mistake: A father cannot pass an X-linked allele to his sons, because sons receive his Y. Always write sex-linked alleles as superscripts on an uppercase X.

Pedigree charts Spec D3.2.13

In a **pedigree chart**, squares are males and circles are females, shaded if affected. A horizontal line joins parents, and a vertical line leads down to their children. Generations are numbered with Roman numerals (I, II, III) and individuals with numbers.

Clues to the pattern of inheritance

Pattern	Clues in the pedigree
Autosomal recessive	Two unaffected parents have an affected child, so both parents must be carriers; males and females affected equally
Autosomal dominant	Every affected individual has at least one affected parent; two affected parents can have an unaffected child
Sex-linked (X-linked) recessive	Mostly males affected, usually with unaffected carrier mothers; an affected father never passes it to his sons



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A pedigree for an **autosomal recessive** condition. Unaffected parents I-1 and I-2 have an affected child, so both must be carriers.

Inductive and deductive reasoning

Working out the pattern from part of a pedigree is **inductive reasoning**: a general conclusion (a theory) from some but not all cases. Using that theory to work out the genotypes of specific individuals is **deductive reasoning**.

Marriage between close relatives

Close relatives share many alleles inherited from common ancestors. If both partners carry the **same rare recessive allele**, each child has a 0.25 chance of being homozygous recessive and affected. This is the genetic basis for the prohibition of marriage between close relatives in many societies.

Exam tip: When deducing a pattern, quote the individuals: for example, 'II-1 is affected but I-1 and I-2 are not, so the allele is recessive'.

Continuous variation and box-and-whisker plots Spec D3.2.14, D3.2.15

Polygenic inheritance is the control of one trait by **many genes**, each with a small additive effect. With many genes the number of possible genotypes is large, so phenotypes form a continuous range, usually close to a normal distribution. Environmental factors add further variation.

- **Skin colour** in humans is polygenic: several genes affect the amount and type of melanin. Environment also matters, because exposure to sunlight increases melanin production.
- A **continuous variable**, such as skin colour or height, can take any value in a range.
- A **discrete variable**, such as ABO blood group, falls into distinct categories with no intermediates.

Mean	The sum of all values divided by the number of values.
Median	The middle value when the data are in order.
Mode	The most frequent value (or class).

Box-and-whisker plots

A **box-and-whisker plot** displays six aspects of a data set: **outliers, minimum, first quartile (Q1), median, third quartile (Q3) and maximum**. The box runs from Q1 to Q3 with a line at the median. The whiskers reach the lowest and highest values that are not outliers.

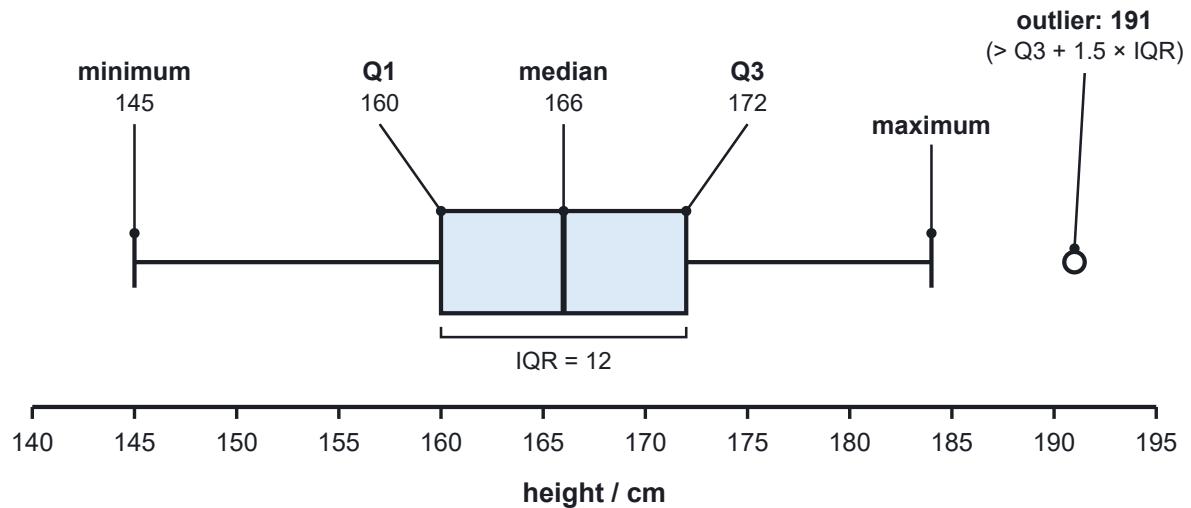
interquartile range (IQR) = $Q3 - Q1$; outlier if $> Q3 + 1.5 \times IQR$ or $< Q1 - 1.5 \times IQR$

Outliers in student height data

Heights of a class of students (cm): $Q1 = 160$, median = 166, $Q3 = 172$. The shortest student is 145 cm and the tallest is 191 cm. Determine whether either is an outlier.

1. $IQR = 172 - 160 = 12$ cm.
2. $1.5 \times IQR = 18$ cm.
3. Upper limit = $172 + 18 = 190$ cm. Lower limit = $160 - 18 = 142$ cm.
4. $191 \text{ cm} > 190 \text{ cm}$, so it is an outlier. $145 \text{ cm} > 142 \text{ cm}$, so it is not.

Answer: 191 cm is an outlier (plotted as a separate point); the upper whisker ends at the tallest non-outlier. 145 cm is the minimum.



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The six aspects of a **box-and-whisker plot**, using the student height data from the worked example. 191 cm is beyond 190 cm ($Q3 + 1.5 \times IQR$), so it is plotted as an outlier.

Maths skill: Box plots let you compare two samples quickly: compare medians for the typical value and IQRs for spread. State the units every time.

Exam tip: Linking question: what are the principles of effective sampling in biological research? A larger random sample gives quartiles and medians that better represent the population.

Quick check

1. Why do AA and Aa individuals have the same phenotype?
2. Give the genotype of a person with blood group O.
3. What phenotype does a heterozygote show in incomplete dominance?
4. State the outlier rule for a box-and-whisker plot.

3 Homeostasis Spec D3.3

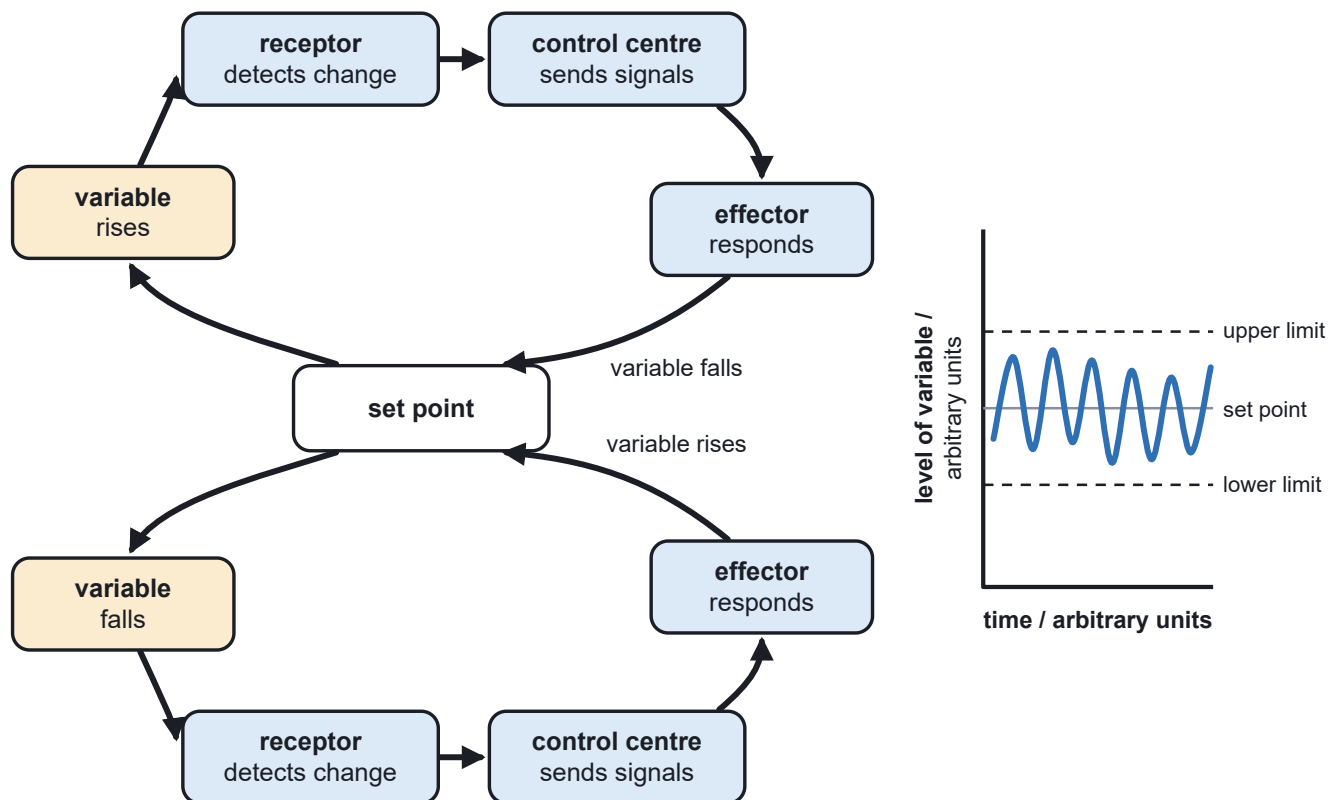
Homeostasis and negative feedback Spec D3.3.1, D3.3.2

Homeostasis is the maintenance of the internal environment of an organism. Variables are **kept within preset limits**, despite fluctuations in the external environment, so that cells, and especially enzymes, work reliably.

Homeostatic variables in humans	
Variable	Approximate normal value
Body (core) temperature	About 37 °C
Blood pH	7.35–7.45
Blood glucose concentration	About 4–6 mmol L ⁻¹ when fasting
Blood osmotic concentration	About 0.3 osmol L ⁻¹

Negative feedback loops

1. A **receptor** detects a change in a variable away from its **set point**.
 2. A **control centre** (often in the brain or an endocrine gland) compares it with the set point and sends signals by nerves or hormones.
 3. **Effectors** (muscles or glands) respond.
 4. The response **reverses the change**, returning the variable to the set point. The loop then switches off.
- Negative feedback works in **both directions**: it brings a variable down when it is above the set point and up when it is below it.
 - Positive feedback amplifies a change away from the set point, so it would make an imbalance worse. That is why homeostasis uses **negative feedback**; positive feedback is only useful for events that must be completed quickly, such as childbirth.



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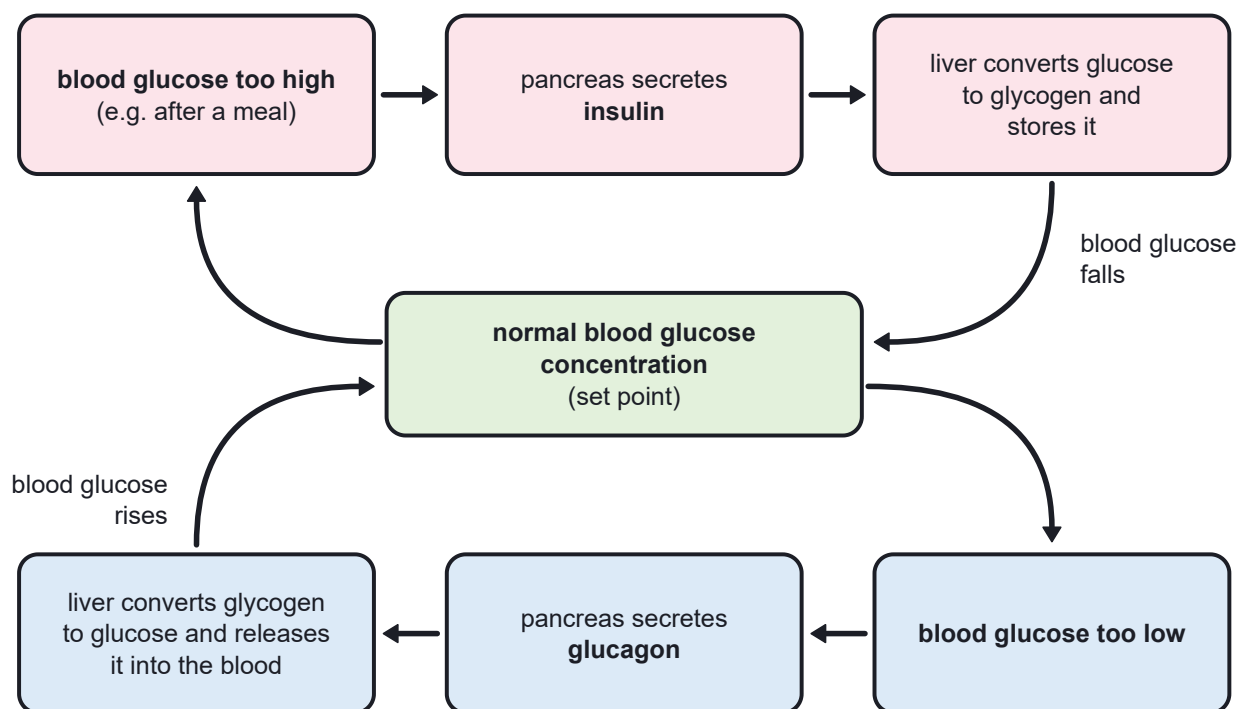
Negative feedback corrects changes in both directions, so the variable fluctuates narrowly around the set point within preset limits.

Exam tip: Negative feedback does not keep a variable perfectly constant: it fluctuates slightly around the set point within preset limits.

Regulation of blood glucose Spec D3.3.3

The pancreas contains clusters of endocrine cells called **islets of Langerhans**. They secrete hormones directly into blood capillaries, which carry them around the body to **target cells** with the right receptors.

Insulin and glucagon		
	Insulin	Glucagon
Secreted by	β (beta) cells of the islets	α (alpha) cells of the islets
Stimulus	Blood glucose above the set point (e.g. after a meal)	Blood glucose below the set point (e.g. fasting, exercise)
Target cells	Liver, skeletal muscle and adipose cells	Mainly liver cells (hepatocytes)
Effects	More glucose transporters inserted into membranes, so cells absorb glucose; glucose converted to glycogen (and fat); more glucose used in respiration	Glycogen broken down to glucose, which is released into the blood; glucose also made from other compounds
Result	Blood glucose falls	Blood glucose rises



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Blood glucose is controlled by **negative feedback**: insulin lowers it and glucagon raises it. The figure shows the liver; insulin also makes skeletal muscle and adipose cells absorb glucose.

Common mistake: Glucagon, glycogen and glucose are easily confused. Glucagon is the hormone; glycogen is the storage polysaccharide; glucose is the sugar in the blood.

Type 1 and type 2 diabetes Spec D3.3.4

Comparing type 1 and type 2 diabetes		
	Type 1	Type 2
Physiological change	Autoimmune destruction of the β cells, so little or no insulin is secreted	Target cells become less responsive to insulin (insulin resistance), e.g. fewer working insulin receptors or glucose transporters; insulin is still produced, though output may fall later
Usual onset	Often in childhood or adolescence	Usually in adults, increasingly in younger people
Risk factors	Genetic predisposition; possibly environmental triggers such as viral infection	Obesity, physical inactivity, diets high in sugar and fat, increasing age, family history (genetic factors) and ethnicity
Prevention	No known method	Healthy diet, regular exercise and keeping a healthy body mass
Treatment	Regular insulin injections or an insulin pump, matched to blood glucose monitoring, meals and activity	Diet low in sugars and refined carbohydrates, exercise, weight loss; drugs that improve insulin sensitivity; insulin in some cases

In both types, blood glucose rises after meals and stays high (**hyperglycaemia**). Glucose appears in the urine and, over many years, high glucose damages blood vessels, nerves, kidneys and the retina.

Exam tip: For 'distinguish' questions, state the difference in physiology directly: type 1, no insulin is produced; type 2, insulin is produced but target cells do not respond to it.

Thermoregulation Spec D3.3.5, D3.3.6

Birds and mammals keep a constant body temperature by both **physiological** and **behavioural** means (for example seeking shade, huddling, or in humans putting on clothes). The details below are for humans.

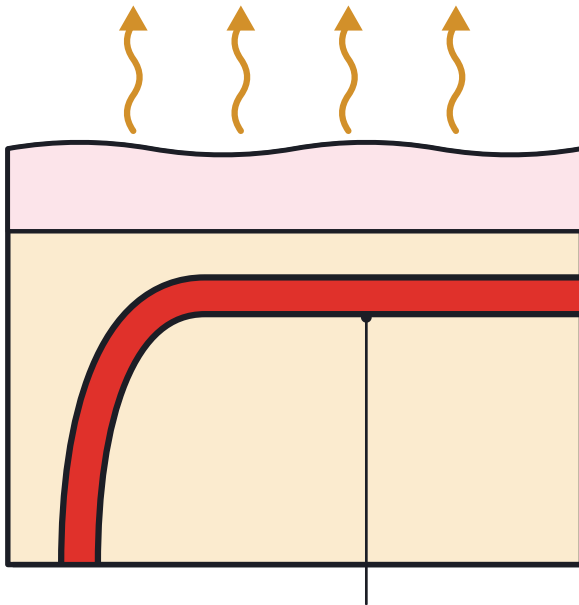
The feedback loop

- **Receptors:** **peripheral thermoreceptors** in the skin detect skin temperature; thermoreceptors in the hypothalamus detect blood (core) temperature.
- **Control centre:** the **hypothalamus** compares temperature with the set point (about 37 °C) and sends nerve impulses to effectors.
- **Hormonal route:** when cold, the hypothalamus stimulates the pituitary gland to secrete TSH, which stimulates the thyroid gland to secrete **thyroxine**. Thyroxine raises the basal metabolic rate, so cells generate more heat.
- **Effectors:** skeletal muscle, brown adipose tissue, skin arterioles, sweat glands and hair erector muscles.

Thermoregulation mechanisms in humans		
Mechanism	When	How it works
Vasoconstriction	Too cold	Arterioles supplying skin capillaries narrow, so less blood flows near the surface and less heat is lost
Shivering	Too cold	Rapid involuntary contractions of skeletal muscle; respiration releases heat
Uncoupled respiration in brown adipose tissue	Too cold	Brown fat cells have many mitochondria with a protein (thermogenin) that lets protons leak back across the inner membrane, so energy is released as heat instead of making ATP
Hair erection	Too cold	Hair erector muscles raise body hairs, trapping a layer of air; much less effective in humans than in furry mammals
Vasodilation	Too hot	Arterioles supplying skin capillaries widen, so more blood flows near the surface and more heat is lost by radiation
Sweating	Too hot	Sweat glands secrete sweat; evaporation of water from the skin removes heat

Too hot: vasodilation

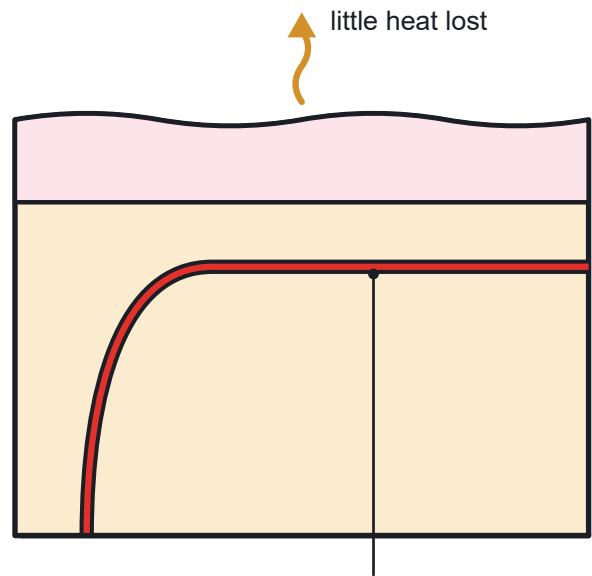
lots of heat lost



Blood vessel widens
more blood near surface

Too cold: vasoconstriction

little heat lost



Blood vessel narrows
less blood near surface

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Vasodilation and **vasoconstriction** of the arterioles supplying skin capillaries change how much blood flows near the skin surface. The vessels widen or narrow; they do not move.

Common mistake: Capillaries do not move up or down in the skin, and they cannot constrict. It is the arterioles supplying them that dilate or constrict.

Exam tip: Linking question: what biological systems are sensitive to temperature changes? Enzyme activity, membrane fluidity and nerve conduction all change with temperature.

Quick check

1. Name four homeostatic variables in humans.
2. Which pancreatic cells secrete glucagon?
3. State the physiological cause of type 1 diabetes.
4. How does brown adipose tissue generate heat?

Quick check answers

Reproduction

1. Offspring are genetically identical to a parent that is already adapted to the existing environment.
2. An LH surge, caused by positive feedback from high oestradiol levels.
3. They are destroyed; only the sperm nucleus contributes.
4. A sticky surface that receives pollen.

Inheritance

1. One dominant allele produces enough functional protein for the trait to appear.
2. ii.
3. An intermediate phenotype, e.g. pink four o'clock flowers.
4. More than $1.5 \times \text{IQR}$ above $Q3$ or below $Q1$.

Homeostasis

1. Body temperature, blood pH, blood glucose concentration and blood osmotic concentration.
2. α (alpha) cells of the islets of Langerhans.
3. Autoimmune destruction of the β cells, so little or no insulin is secreted.
4. Uncoupled respiration: energy is released as heat instead of being used to make ATP.